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FILE 'HOME' ENTERED AT 22:50:22 ON 10 JUN 2004

=> file biosis
COST IN U.S. DOLLARS                                    SINCE FILE      TOTAL
                                                           ENTRY    SESSION
FULL ESTIMATED COST                                         0.21       0.21

FILE 'BIOSIS' ENTERED AT 22:50:38 ON 10 JUN 2004
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FILE COVERS 1969 TO DATE.
CAS REGISTRY NUMBERS AND CHEMICAL NAMES (CNs) PRESENT
FROM JANUARY 1969 TO DATE.

RECORDS LAST ADDED: 9 June 2004 (20040609/ED)

FILE RELOADED: 19 October 2003.

=> s (smith (w) lemli (w) optiz)
         10498 SMITH
            78 SMITHS
         10555 SMITH
                  (SMITH OR SMITHS)
           563 LEMLI
            23 OPTIZ
L1           16 (SMITH (W) LEMLI (W) OPTIZ)

=> d l1 1-16 ibib,iabs

L1   ANSWER 1 OF 16  BIOSIS  COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER:    2004:119409  BIOSIS
DOCUMENT NUMBER:     PREV200400121822
TITLE:               Rod photoreceptor responses in children with
                     Smith-Lemli-Opitz syndrome.
AUTHOR(S):           Elias, Ellen R.; Hansen, Ronald M.; Irons, Mira; Quinn,
                     Nicole B.; Fulton, Anne B. [Reprint Author]
CORPORATE SOURCE:    300 Longwood Ave, Boston, MA, 02115, USA
                     anne.fulton@tch.harvard.edu
SOURCE:              Archives of Ophthalmology, (December 2003) Vol. 121, No.
                     12, pp. 1738-1743. print.
                     ISSN: 0003-9950 (ISSN print).
DOCUMENT TYPE:       Article
LANGUAGE:            English
ENTRY DATE:          Entered STN: 3 Mar 2004
                     Last Updated on STN: 3 Mar 2004
ABSTRACT:
Objective: To test the hypothesis that the kinetics of activation and
deactivation of rod phototransduction are altered in children with
Smith-Lemli-Opitz syndrome (SLOS), a common genetic disorder caused by an
inborn error in cholesterol biosynthesis.
Methods: Thirteen patients with SLOS (median age, 4 years) were studied by
means of scotopic full-field electroretinography.
The kinetics of activation and deactivation of rod phototransduction were
derived from the electroretinographic a-wave.
Postreceptoral electroretinographic components were also evaluated.
Results: The kinetics of activation were below normal limits in all but 3 of
the 13 patients.
Rod cell recovery (deactivation) in SLOS was slower than normal in all 8
patients in whom it was studied.
Postreceptoral sensitivity was below normal limits in all but 1 of the 13
patients.
Conclusions: The kinetics of phototransduction are slow in children with SLOS.
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This is likely a consequence of altered sterol composition in the cell membranes of the rod photoreceptors.
To our knowledge, this is the first demonstration of altered kinetics of a membrane-bound signaling system in SLOS.
Investigation of other membrane-bound signaling systems may be warranted in the quest to understand development and phenotype of individuals with SLOS.

L1 ANSWER 2 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2002:65533 BIOSIS
DOCUMENT NUMBER: PREV200200065533
TITLE: Rapid screening test for **Smith-Lemli-Optiz** syndrome.
AUTHOR(S): Hercules, D. M. [Inventor]; Naylor, E. W. [Inventor]; Zimmerman, P. A. [Inventor]
CORPORATE SOURCE: Nashville, Tenn., USA
ASSIGNEE: UNIVERSITY OF PITTSBURGH OF THE COMMONWEALTH SYSTEM OF HIGHER EDUCATION
PATENT INFORMATION: US 5629210 May 13, 1997
SOURCE: Official Gazette of the United States Patent and Trademark Office Patents, (May 13, 1997) Vol. 1198, No. 2, pp. 1124. print.
CODEN: OGUPE7. ISSN: 0098-1133.
DOCUMENT TYPE: Patent
LANGUAGE: English
ENTRY DATE: Entered STN: 9 Jan 2002
Last Updated on STN: 25 Feb 2002

L1 ANSWER 3 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2000:87714 BIOSIS
DOCUMENT NUMBER: PREV200000087714
TITLE: Synthesis of (3alpha-3H)cholesta-5,8-dien-3beta-ol and tritium-labeled forms of other sterols of potential importance in the **Smith-Lemli-Optiz** syndrome.
AUTHOR(S): Ruan, Benfang; Wilson, William K. [Reprint author]; Pang, Jihai; Schroepfer, George J., Jr.
CORPORATE SOURCE: Department of Biochemistry and Cell Biology, Rice University, 6100 Main St., Houston, TX, 77005-1892, USA
SOURCE: Steroids, (Jan., 2000) Vol. 65, No. 1, pp. 29-39. print.
CODEN: STEDAM. ISSN: 0039-128X.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 10 Mar 2000
Last Updated on STN: 3 Jan 2002
ABSTRACT:
Five unsaturated sterols relevant to the Smith-Lemli-Opitz syndrome have been prepared in high radiochemical purity with a tritium label at the 3alpha position.
Swern oxidation of cholesta-5,8-dien-3beta-ol and other unlabeled C27 sterols afforded the corresponding 3-ketosteroids, and reduction with tritiated NaBH4 gave the desired 3alpha-3H sterols, with double bonds at the DELTA5,8, DELTA5,8(14), DELTA6,8, DELTA6,8(14), and DELTA8 positions.
High radiochemical purity of the tritiated sterols was demonstrated by normal phase, reversed phase, and silver-ion (Ag+) high-performance liquid chromatography (HPLC).
In the course of this work, we developed a medium-pressure variant of Ag+-HPLC for purifying radiolabeled samples, documented significant isotopic fractionation of the 3alpha-tritiated sterols and their acetates on Ag+-HPLC, and discovered unexpected effects of a DELTA8(14) bond on the conformation of 3-keto-DELTA5-steroids.
The synthetic and analytical methodologies described herein should provide a sound basis for investigating the origin and metabolism of sterols involved in the Smith-Lemli-Opitz syndrome and in late stages of cholesterol biosynthesis.

L1 ANSWER 4 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN

ACCESSION NUMBER: 1999:449378 BIOSIS
DOCUMENT NUMBER: PREV199900449378
TITLE: Carbon recycling into de novo lipogenesis is a major pathway in neonatal metabolism of linoleate and alpha-linolenate.
AUTHOR(S): Cunnane, S. C. [Reprint author]; Menard, C. R.; Likhodii, S. S.; Brenna, J. T.; Crawford, M. A.
CORPORATE SOURCE: Department of Nutritional Sciences, University of Toronto, Toronto, ON, M5S 3E2, Canada
SOURCE: Prostaglandins Leukotrienes and Essential Fatty Acids, (May-June, 1999) Vol. 60, No. 5-6, pp. 387-392. print.
CODEN: PLEAEU. ISSN: 0952-3278.
DOCUMENT TYPE: Article
General Review; (Literature Review)
LANGUAGE: English
ENTRY DATE: Entered STN: 26 Oct 1999
Last Updated on STN: 26 Oct 1999

ABSTRACT:
Recent reports indicate that recycling of the beta-oxidized carbon skeleton of linoleate and alpha-linolenate into newly synthesized cholesterol and fatty acids in the brain is quantitatively significant in both suckling rats and pre- and postnatally in rhesus monkeys.
The recycling appears to occur via ketones which are not only readily produced from these 18 carbon polyunsaturates but are also the main lipogenic precursors for the developing mammalian brain.
Since the neonatal rat brain appears not to acquire cholesterol or long chain saturated or monounsaturated fatty acids from the circulation, ketones and ketogenic precursors seem to be crucial for normal brain synthesis of these lipids.
Cholesterol is plentiful in brain membranes and it has also been discovered to be the essential lipid adduct of the 'hedgehog' family of proteins, the appropriate expression of which determines normal embryonic tissue patterning and neurological development.
Insufficient cholesterol or inappropriate expression of 'sonic hedgehog' has major adverse neurodevelopmental consequences typified in humans by ***Smith*** -**Lemli-Optiz** syndrome.
Hence, we propose that the importance of alpha-linolenate and linoleate for normal neural development arises not only from being precursors to longer chain polyunsaturates incorporated into neuronal membranes but, perhaps equally importantly, by being ketogenic precursors needed for in situ brain lipid synthesis.

L1 ANSWER 5 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1999:389675 BIOSIS
DOCUMENT NUMBER: PREV199900389675
TITLE: Antenatal therapy of **Smith-Lemli-Optiz** syndrome.
AUTHOR(S): Irons, Mira B. [Reprint author]; Nores, Jose; Stewart, Theresa L.; Craigo, Sabrina D.; Bianchi, Diana W.; D'Alton, Mary E.; Tint, G. Stephen; Salen, Gerald; Bradley, Linda A.
CORPORATE SOURCE: Fegan 10, Division of Genetics and Metabolism, Children's Hospital, 300 Longwood Avenue, Boston, MA, 02115, USA
SOURCE: Fetal Diagnosis and Therapy, (May-June, 1999) Vol. 14, No. 3, pp. 133-137. print.
ISSN: 1015-3837.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 28 Sep 1999
Last Updated on STN: 28 Sep 1999

ABSTRACT:
Objectives: **Smith-Lemli-Optiz** syndrome (SLOS) is a recessively inherited disorder caused by an inborn error of cholesterol metabolism that results in deficiency of cholesterol and accumulation of the cholesterol precursor, 7-dehydrocholesterol (DHC) and its epimer, 8-DHC. Affected patients present with congenital anomalies, growth restriction, and

mental retardation.
Postnatal treatment with cholesterol supplementation has been shown to improve plasma sterol levels and has resulted in improved growth and development in many patients.
We hypothesized that prenatal supplementation of cholesterol could potentially arrest some of the adverse consequences of cholesterol deficiency at an earlier stage of development.
Methods: SLOS was diagnosed in the third trimester in a fetus initially identified by sonography with intrauterine growth restriction and ambiguous genitalia and confirmed by elevated levels of 7- and 8-DHC in amniotic fluid. Antenatal supplementation of cholesterol was provided by fetal intravenous and intraperitoneal transfusions of fresh frozen plasma (cholesterol level = 219 mg/dl).
Results: The in utero transfusions resulted in increased levels of fetal cholesterol, as measured in blood samples obtained by cordocentesis. In addition, fetal red cell mean corpuscular volume rose, which further indicated that the exogenous cholesterol was incorporated into the fetal erythrocytes.
Conclusions: Antenatal treatment of SLOS by cholesterol supplementation is feasible and results in improvement in fetal plasma cholesterol levels and fetal red cell volume.
SLOS may be added to the growing list of human genetic disorders for which prenatal diagnosis is available and therapeutic intervention may be possible.

L1 ANSWER 6 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1999:129782 BIOSIS
DOCUMENT NUMBER: PREV199900129782
TITLE: Cholesterol supplementation does not alter development progress in **Smith-Lemli-Optiz** syndrome.
AUTHOR(S): Ruggiero, M. [Reprint author]; Quiggens, D.; Petit-Kekel, K.; Linck, L.; Steiner, R.
CORPORATE SOURCE: Dep. Molecular, Doernbecker Children's Hosp., Child. Dev. Rehab. Cent., Portland, OR, USA
SOURCE: Journal of Investigative Medicine, (Feb., 1999) Vol. 47, No. 2, pp. 56A. print.
Meeting Info.: Western Regional Meeting of the American Federation for Medical Research. Carmel, California, USA. January 27-30, 1999. American Federation for Medical Research.
ISSN: 1081-5589.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 17 Mar 1999
Last Updated on STN: 17 Mar 1999

L1 ANSWER 7 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1999:54980 BIOSIS
DOCUMENT NUMBER: PREV199900054980
TITLE: Eye findings in 8 children and a spontaneously aborted fetus with RSH/**Smith-Lemli-Optiz** syndrome.
AUTHOR(S): Atchaneeyasakul, La-Ongsri; Linck, Leesa M.; Connor, William E.; Weleber, Richard G.; Steiner, Robert D. [Reprint author]
CORPORATE SOURCE: Dep. Pediatr., Oregon Health Sci. Univ., 3181 S.W. Sam Jackson Park Road, Portland, OR 97201-3098, USA
SOURCE: American Journal of Medical Genetics, (Dec. 28, 1998) Vol. 80, No. 5, pp. 501-505. print.
ISSN: 0148-7299.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 16 Feb 1999
Last Updated on STN: 16 Feb 1999

ABSTRACT:
We evaluate the ophthalmologic findings in 8 children with RSH/Smith-Lemli-Opitz syndrome (SLOS) and document abnormal concentrations of cholesterol and cholesterol precursors in the ocular tissues in a case of SLOS.
The most common ophthalmologic finding was blepharoptosis, which was found in 6 of 8 patients, with the severity ranging from mild to moderate.
None of the patients in the present study demonstrated cataracts; none had amblyopia from blepharoptosis.
One patient had a right hypertropia with overaction of the inferior oblique muscle.
This patient also had optic atrophy and a second patient had bilateral optic nerve hypoplasia.
The importance of these findings to the visual function remains to be defined.
Sterol analysis from ocular tissues of an aborted fetus with SLOS showed increased 7- and 8-dehydrocholesterol and a low cholesterol concentration in the retinal pigment epithelium, lens, cornea, and sclera.
Routine ophthalmologic examination is indicated in SLOS because of the high incidence of abnormalities, most likely due to the abnormal synthesis of cholesterol and cholesterol precursors in the ocular tissues of these patients, as evidenced by sterol analysis of the ocular tissues in a case of SLOS.

L1 ANSWER 8 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1998:376001 BIOSIS
DOCUMENT NUMBER: PREV199800376001
TITLE: Photosensitivity in the **Smith Lemli Optiz** syndrome: Correlation with levels of 7-dehydrocholesterol.
AUTHOR(S): Anstey, A. [Reprint author]; Pearce, A. D.; Venn, C.; Ryan, A.
CORPORATE SOURCE: Dep. Dermatol., Royal Gwent Hosp., Newport, UK
SOURCE: British Journal of Dermatology, (April, 1998) Vol. 138, No. 4, pp. 731. print.
Meeting Info.: Annual Meeting of the British Society for Investigative Dermatology and British Photodermatology Group. Liverpool, England, UK. April 22-24, 1998. British Photodermatology Group; British Society for Investigative Dermatology.
CODEN: BJDEAZ. ISSN: 0007-0963.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 2 Sep 1998
Last Updated on STN: 2 Sep 1998

L1 ANSWER 9 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1998:159639 BIOSIS
DOCUMENT NUMBER: PREV199800159639
TITLE: Toxicity in embryo culture of cholesterol precursor and oxidized by-products accumulated in **Smith-Lemli-Optiz**-syndrome.
AUTHOR(S): Gaoua, W. [Reprint author]; Wolf, C.; Chevy, F.; Haddadi, K.; Citadelle, D.; Mulliez, N.; Roux, C. [Reprint author]
CORPORATE SOURCE: Lab. Embryologie, CHU St.-Antoine, Paris, France
SOURCE: Teratology, (Dec., 1997) Vol. 56, No. 6, pp. 398. print.
Meeting Info.: 25th Conference of the European Teratology Society. Cannes, France. September 14-17, 1997. European Teratology Society.
CODEN: TJADAB. ISSN: 0040-3709.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
Conference; (Meeting Poster)
LANGUAGE: English
ENTRY DATE: Entered STN: 31 Mar 1998
Last Updated on STN: 4 May 1998

L1 ANSWER 10 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1998:110017 BIOSIS
DOCUMENT NUMBER: PREV199800110017
TITLE: Prenatal diagnosis of the Smith-Lemli-Opitz syndrome (SLOS) by the analysis of amniotic fluid (AF) sterols is accurate and precise.
AUTHOR(S): Tint, G. S. [Reprint author]; Abuelo, D.; Irons, M.; Honda, A.; Honda, M.; Salen, G. [Reprint author]
CORPORATE SOURCE: VA Med. Cent., East Orange, NJ, USA
SOURCE: American Journal of Human Genetics, (Oct., 1997) Vol. 61, No. 4 SUPPL., pp. A19. print.
Meeting Info.: 47th Annual Meeting of the American Society of Human Genetics. Baltimore, Maryland, USA. October 28-November 1, 1997.
CODEN: AJHGAG. ISSN: 0002-9297.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
Conference; (Meeting Slide)
LANGUAGE: English
ENTRY DATE: Entered STN: 3 Mar 1998
Last Updated on STN: 3 Mar 1998

L1 ANSWER 11 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1994:270790 BIOSIS
DOCUMENT NUMBER: PREV199497283790
TITLE: **Smith-Lemli-Optiz** syndrome in a female with a de novo, balanced translocation involving 7q32: Probable disruption of an SLOS gene.
AUTHOR(S): Wallace, Margaret [Reprint author]; Zori, Roberto T.; Alley, Tiffany; Whidden, Elaine; Gray, Brian A.; Williams, Charles A.
CORPORATE SOURCE: Pediatric Genetics, University Florida, PO Box 100296, HSC, Gainesvill, FL 32610-0296, USA
SOURCE: American Journal of Medical Genetics, (1994) Vol. 50, No. 4, pp. 368-374.
ISSN: 0148-7299.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 24 Jun 1994
Last Updated on STN: 25 Jun 1994

L1 ANSWER 12 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1990:197599 BIOSIS
DOCUMENT NUMBER: PREV199089104270; BA89:104270
TITLE: APPARENT SMITH-LEMLI-OPITZ SYNDROME IN A CHILD WITH A PREVIOUSLY UNDESCRIBED FORM OF MUCOLIPIDOSIS NOT INVOLVING THE NEURONS.
AUTHOR(S): PARNES S [Reprint author]; HUNTER A G W; JIMENEZ C; CARPENTER B F; MACDONALD I
CORPORATE SOURCE: DIV GENETICS, CHILD HOSP EASTERN ONTARIO, 401 SMYTH ROAD, OTTAWA, ONTARIO, CAN K1H 8L1
SOURCE: American Journal of Medical Genetics, (1990) Vol. 35, No. 3, pp. 397-405.
ISSN: 0148-7299.
DOCUMENT TYPE: Article
FILE SEGMENT: BA
LANGUAGE: ENGLISH
ENTRY DATE: Entered STN: 24 Apr 1990
Last Updated on STN: 24 Apr 1990
ABSTRACT:
A diagnosis of Smith-Lemli-Opitz syndrome was made shortly after birth in a small-for-dates infant, on the basis of a characteristic face, penoscrotal hypospadias, bilateral postaxial hexadactyly, and bilateral syndactyly of toes 2-3.
The clinical course was marked by failure to thrive, severe delay, refractory

myoclonic jerks beginning at age 2 months, and increasing hepatosplenomegaly. He developed corneal clouding and increased gingival hypertrophy and died at age 18 weeks.
Autopsy disclosed widespread storage of mucopolysaccharides and lipids within the macrophages and, to a lesser extent, parenchymal cells, of all organ systems.
There was extensive demyelination of the cerebral white matter, and dystrophic calcification in the cerebrum, cerebellum and brainstem.
There was no evidence of primary neuronal involvement in the storage.
Although the chance concurrence of 2 uncommon diseases is rare, a causal link between the clinical anomalies and the storage disorder cannot be argued convincingly on the basis of one case.
Careful pathologic studies of other children who die with clinical signs compatible with **Smith-Lemli-Optiz** syndrome are indicated.

L1 ANSWER 13 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1987:145294 BIOSIS
DOCUMENT NUMBER: PREV198783074344; BA83:74344
TITLE: POSSIBLE ABNORMALITIES OF STEROID SECRETION IN CHILDREN WITH SMITH-LEMLI-OPITZ SYNDROME AND THEIR PARENTS.
AUTHOR(S): CHASALOW F I [Reprint author]; BLETHEN S L; TAYSI K
CORPORATE SOURCE: DIV PEDIATR ENDOCRINOL, SCHNEIDER CHILDREN'S HOSP LONG ISLAND JEWISH MED CENT, NEW HYDE PARK, NY 11042, USA
SOURCE: Steroids, (1985) Vol. 46, No. 4-5, pp. 827-844.
CODEN: STEDAM. ISSN: 0039-128X.
DOCUMENT TYPE: Article
FILE SEGMENT: BA
LANGUAGE: ENGLISH
ENTRY DATE: Entered STN: 21 Mar 1987
Last Updated on STN: 21 Mar 1987
ABSTRACT:
In early infancy, two unrelated children with **Smith-Lemli-** ***Optiz*** syndrome were found to have elevated levels of androgen sulfates. When the steroid conjugates in the serum of normal infants were hydrolyzed and chromatographed on Sephadex LH-20, 4 androgen containing peaks (I, II, III, IV) were found.
In the serum from these two infants with Smith-Lemli-Opitz syndrome, Peaks I and III were increased, but Peaks II and IV were absent.
The parents of the two children, and of three additional unrelated children with Smith-Lemli-Opitz syndrome, had exaggerated 17-hydroxyprogesterone responses to an intravenous bolus of ACTH.
These findings suggest that a defect in steroid metabolism may be linked to the ***Smith*** **-Lemli-Optiz** syndrome.

L1 ANSWER 14 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1987:60828 BIOSIS
DOCUMENT NUMBER: PREV198732031049; BR32:31049
TITLE: MULTIPLE MALFORMATION SYNDROME SUGGESTIVE OF **SMITH -LEMLI-OPTIZ** SYNDROME WITH HOLOPROSENCEPHALY IN A 46 XY STILLBORN INFANT.
AUTHOR(S): LIEBER E [Reprint author]; VALDARAMA E; CASALOW F
CORPORATE SOURCE: SCHNEIDER CHILDREN'S HOSPITAL OF LONG ISLAND JEWISH MED CENTER, NEW HYDE PARK, NEW YORK, USA
SOURCE: American Journal of Human Genetics, (1986) Vol. 39, No. 3 SUPPL, pp. A69.
Meeting Info.: 37TH ANNUAL MEETING OF THE AMERICAN SOCIETY OF HUMAN GENETICS, PHILADELPHIA, PA., USA, NOV. 2-5, 1986. AM J HUM GENET.
CODEN: AJHGAG. ISSN: 0002-9297.
DOCUMENT TYPE: Conference; (Meeting)
FILE SEGMENT: BR
LANGUAGE: ENGLISH
ENTRY DATE: Entered STN: 17 Jan 1987
Last Updated on STN: 17 Jan 1987

L1 ANSWER 15 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1986:258713 BIOSIS
DOCUMENT NUMBER: PREV198682013462; BA82:13462
TITLE: AN AUTOSOMAL RECESSIVE MENTAL RETARDATION SYNDROME WITH HEPATIC FIBROSIS AND RENAL CYSTS.
AUTHOR(S): THOMPSON E [Reprint author]; BARAITSER M
CORPORATE SOURCE: CLIN GENETICS UNIT, HOSP SICK CHILDREN, GREAT ORMOND ST, LONDON WC1N 3JH, ENGL, UK
SOURCE: American Journal of Medical Genetics, (1986) Vol. 24, No. 1, pp. 151-158.
ISSN: 0148-7299.
DOCUMENT TYPE: Article
FILE SEGMENT: BA
LANGUAGE: ENGLISH
ENTRY DATE: Entered STN: 21 Jun 1986
Last Updated on STN: 21 Jun 1986

ABSTRACT:
Two sisters had developmental retardation and congenital hepatic fibrosis. One, 23 years old, had facial anomalies reminiscent of Smith-Lemli-Opitz syndrome, ocular coloboma, and hypoplastic kidneys with a single cyst. The other sister died at 18 months and had an encephalocele and cystically dilated collecting ducts in the renal medulla. Although the manifestations in these two sisters are similar to the ***Smith*** -**Lemli**-**Optiz** and Meckel syndromes respectively, there are sufficient differences to suggest that they had a separate autosomal recessive MCA-MR syndrome.

L1 ANSWER 16 OF 16 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1973:119618 BIOSIS
DOCUMENT NUMBER: PREV197355019611; BA55:19611
TITLE: **SMITH LEMLI OPTIZ** SYNDROME IN AN INBRED FAMILY.
AUTHOR(S): SARA N; BENDERLY A; LEVY J; KATZNELSON M B-M
SOURCE: American Journal of Diseases of Children, (1972) Vol. 124, No. 3, pp. 431-433.
CODEN: AJDCAI. ISSN: 0002-922X.
DOCUMENT TYPE: Article
FILE SEGMENT: BA
LANGUAGE: Unavailable

=> file caplus
COST IN U.S. DOLLARS

	SINCE FILE ENTRY	TOTAL SESSION
FULL ESTIMATED COST	35.50	35.71

FILE 'CAPLUS' ENTERED AT 22:54:07 ON 10 JUN 2004
USE IS SUBJECT TO THE TERMS OF YOUR STN CUSTOMER AGREEMENT.
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FILE COVERS 1907 - 10 Jun 2004 VOL 140 ISS 24
FILE LAST UPDATED: 9 Jun 2004 (20040609/ED)

This file contains CAS Registry Numbers for easy and accurate substance identification.

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=> s (smith (w) lemli (w) optiz)
         11808 SMITH
            29 SMITHS
         11833 SMITH
                 (SMITH OR SMITHS)
           151 LEMLI
             7 OPTIZ
L2           4 (SMITH (W) LEMLI (W) OPTIZ)

=> d l2 1-4 ibib,iabs
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L2 ANSWER 1 OF 4 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 2003:446875 CAPLUS
TITLE: Adrenal insufficiency and abnormal genitalia in a 46XX female with **smith-lemli-optiz** syndrome
AUTHOR(S): Chemaitilly, W.; Goldenberg, A.; Baujat, G.; Thibaud, E.; Cormier-Daire, V.; Abadie, V.
CORPORATE SOURCE: Paediatric Endocrinology Unit, Hopital Necker-Enfants Malades, Paris, Fr.
SOURCE: Hormone Research (2003), 59(5), 254-256
CODEN: HRMRA3; ISSN: 0301-0163
PUBLISHER: S. Karger AG
DOCUMENT TYPE: Journal
LANGUAGE: English
ABSTRACT:

To report the first case of a 46XX female infant with Smith-Lemli-Opitz syndrome (SLOS), adrenal insufficiency and abnormal genitalia. The patient was assessed for hormonal status on day 4 and 6 mo later and was followed-up from the study time (2.5 yr of age). The patient had a persistent urogenital sinus, posterior labial fusion without clitoromegaly. She presented with a salt-wasting syndrome on day 4. Adrenal insufficiency was confirmed. Adrenal androgen levels, including 17-hydroxyprogesterone and 11-deoxycortisol were moderately elevated. Children with SLOS should be assessed for adrenal insufficiency. In female infants, abnormal external genitalia can be observed even if the precise mechanism behind these abnormalities is yet to be determined

REFERENCE COUNT: 7 THERE ARE 7 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L2 ANSWER 2 OF 4 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1999:793625 CAPLUS
DOCUMENT NUMBER: 132:180766
TITLE: Synthesis of [3α-3H]cholesta-5,8-dien-3β-ol and tritium-labeled forms of other sterols of potential importance in the **Smith-Lemli-Optiz** syndrome
AUTHOR(S): Ruan, B.; Wilson, W. K.; Pang, J.; Schroepfer, G. J.
CORPORATE SOURCE: Department of Chemistry, Rice University, Houston, TX, USA
SOURCE: Steroids (2000), 65(1), 29-39
CODEN: STEDAM; ISSN: 0039-128X
PUBLISHER: Elsevier Science Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
ABSTRACT:

Five unsatd. sterols relevant to the Smith-Lemli-Opitz syndrome have been prepared in high radiochem. purity with a tritium label at the 3α position. Swern oxidation of cholesta-5,8-dien-3β-ol and other unlabeled C27 sterols afforded the corresponding 3-ketosteroids, and reduction with tritiated NaBH4 gave

the desired 3α-3H sterols, with double bonds at the Δ5,8, Δ5,8(14), Δ6,8, Δ6,8(14), and Δ8 positions. High radiochem. purity of the tritiated sterols was demonstrated by normal phase, reversed phase, and silver-ion (Ag+) high-performance liquid chromatog. (HPLC). In the course of this work, we developed a medium-pressure variant of Ag+-HPLC for purifying radiolabeled samples, documented significant isotopic fractionation of the 3α-tritiated sterols and their acetates on Ag+-HPLC, and discovered unexpected effects of a Δ8(14) bond on the conformation of 3-keto-Δ5-steroids. The synthetic and anal. methodologies described herein should provide a sound basis for investigating the origin and metabolism of sterols involved in the Smith-Lemli-Opitz syndrome and in late stages of cholesterol biosynthesis.

REFERENCE COUNT: 33 THERE ARE 33 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L2 ANSWER 3 OF 4 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1999:533647 CAPLUS
DOCUMENT NUMBER: 131:269913
TITLE: Carbon recycling into de novo lipogenesis is a major pathway in neonatal metabolism of linoleate and α-linolenate
AUTHOR(S): Cunnane, S. C.; Menard, C. R.; Likhodii, S. S.; Brenna, J. T.; Crawford, M. A.
CORPORATE SOURCE: Department of Nutritional Sciences, Faculty of Medicine, University of Toronto, Toronto, ON, M5S 3E2, Can.
SOURCE: Prostaglandins, Leukotrienes and Essential Fatty Acids (1999), 60(5&6), 387-392
CODEN: PLEAEU; ISSN: 0952-3278
PUBLISHER: Churchill Livingstone
DOCUMENT TYPE: Journal
LANGUAGE: English
ABSTRACT:
Recent reports indicate that recycling of the β-oxidized carbon skeleton of linoleate and α-linolenate into newly synthesized cholesterol and fatty acids in the brain is quant. significant in both suckling rats and pre- and postnatally in rhesus monkeys. The recycling appears to occur via ketones which are not only readily produced from these 18 carbon polyunsaturates but are also the main lipogenic precursors for the developing mammalian brain. Since the neonatal rat brain appears not to acquire cholesterol or long chain saturated or monounsatd. fatty acids from the circulation, ketones and ketogenic precursors seem to be crucial for normal brain synthesis of these lipids. Cholesterol is plentiful in brain membranes and it has also been discovered to be the essential lipid adduct of the "hedgehog" family of proteins, the appropriate expression of which dets. normal embryonic tissue patterning and neurol. development. Insufficient cholesterol or inappropriate expression of "sonic hedgehog" has major adverse neurodevelopmental consequences typified in humans by **Smith-Lemli-Optiz** syndrome. Hence, we propose that the importance of α-linolenate and linoleate for normal neural development arises not only from being precursors to longer chain polyunsaturates incorporated into neuronal membranes but, perhaps equally importantly, by being ketogenic precursors needed for in situ brain lipid synthesis.

REFERENCE COUNT: 52 THERE ARE 52 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

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ACCESSION NUMBER: 1997:722701 CAPLUS
DOCUMENT NUMBER: 128:46816
TITLE: Cholesterol biosynthesis inhibited by BM15.766 induces holoprosencephaly in the rat
AUTHOR(S): Kolf-Clauw, M.; Chevy, F.; Siliart, B.; Wolf, C.; Mulliez, N.; Roux, C.

CORPORATE SOURCE: Toxicology Department, Ecole Nationale Veterinaire d'Alfort, Maisons-Alfort, 94704, Fr.
SOURCE: Teratology (1997), 56(3), 188-200
CODEN: TJADAB; ISSN: 0040-3709
PUBLISHER: Wiley-Liss
DOCUMENT TYPE: Journal
LANGUAGE: English
ABSTRACT:
To confirm that blocking 7-dehydrocholesterol Δ7 reductase (7DHC reductase), as observed in **Smith-Lemli-Optiz** syndrome (SLOS), induces craniofacial defects, the authors tested BM15.766, which blocks 7DHC reductase but is chemical unrelated to the holoprosencephaly-inducing teratogen AY9944. Rats were given BM15.766 either in methylcellulose from days (D) 1 through D11 (3 treated groups: protocol A) or in olive oil from D4 through D7 (300 mg/kg/d: protocol B). The sera were sampled on D0, D3, and D5 or D6, D10, D14, and D21 to measure cholesterol and dehydrocholesterols in all groups and steroid hormones in protocol B. D21 fetuses showed the holoprosencephaly spectrum of malformations and the treated dams low cholesterol and accumulation of 7DHC, 8DHC, and trienols, as in SLOS-affected children. In the 3 dosage groups the malformations were dose-related and enzymic cholesterol decreased to a plateau. The DHC reached 25-44% of the total sterols in the dams. In protocol B, one-third of the BM15.766-treated fetuses presented facial malformations and almost two-thirds pituitary agenesis. On D10, cholesterol reached a min. and the DHC a maximum while estradiol 17β and progesterone were lowered, the latter decreasing in correlation with cholesterolemia. A sterol profile similar to that previously observed after AY9944 associated with a similarly high incidence of pituitary agenesis confirmed that time-limited inhibition of 7DHC reductase induces holoprosencephaly and that pituitary agenesis is the minor form of holoprosencephaly.

REFERENCE COUNT: 61 THERE ARE 61 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

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